Instructions: *Use Times New Roman font size 11, 1.0 pt text space, and* ***maximum three A4 pages****, including any figures.*

Name or type of instrument	**Illumina iScan Dx + Tecan liquid-handling robot**
Unit	Clinical Genomics Umeå
Platform	Clinical Genomics
Applicant (Head of Unit)	Pär Larsson
E-mail address	par.larsson@umu.se
Instrument, acquisition cost (kSEK)	3500 (inkl OH) scanner + approx. 1250 (inkl OH) for Techan Robot
Funding applied for (kSEK)	2375 (50% of purchase ammount inkl OH)

Description of the instrument

Describe the instrument, its purpose, and its specific capabilities. If possible, provide a link to a web page describing the type of instrument. Does the instrument represent a new technology at the unit, or is it a replacement/upgrade of current equipment base? Is the instrument unique in terms of service to users nationally?

The Illumina iScan Dx instrument (https://emea.illumina.com/systems/array-scanners/iscan.html) comprises an array scanner that supports rapid, sensitive, and accurate imaging of Illumina BeadChips for genomic and epigenomic analysis. It uses high-performance lasers, optics, and detection systems to offer a submicron resolution. The instrument is compatible with Tecan system liquid handling robotics that standardizes sample and liquid handling to ensure high quality, reproducibility, and high throughput of samples.

The Clinical Genomics (CG) node in Umeå has a technology focus on Epigenetics and has become recognized for DNA methylation classification of hematological malignancies, with a special focus on childhood T-cell leukemias. CG aims at offering a DNA methylation classification service for hematological malignancies including both laboratory and bioinformatic analysis.

The array-based method for DNA methylation classification using the iScan Dx instrument generates data of high quality at a reasonable cost. The use of DNA methylation as a biomarker has several advantages in clinical diagnostics since DNA is routinely prepared, and can be sent to an external lab for analysis at room temperature.
The Illumina iScan Dx instrument will replace a withdrawn and aging HiScan instrument and ensure that the Umeå node can serve as a national resource for DNA methylation classification of hematological malignancies. DNA methylation has already become integrated into clinical diagnostics for brain tumors, and hematological malignancies are likely to follow.

Statement of need

Describe how the instrument will be crucial for the operations, productivity, development, and competitiveness of your unit. Estimate the need and impact of the instrument from a user perspective. Will the instrument be shared with or used by other units or research groups at SciLifeLab? Also, describe any co-funding plans.

The CG node in Umeå has a good opportunity to become a service platform for methylation classification of hematological malignancies in Sweden and Europe. There is an urgent need for new molecular biomarkers that can improve subtype discrimination, treatment stratification, and prognosis prediction. DNA methylation classification is a promising biomarker in several hematological malignancies, but additional research and clinical trials are needed to support clinical implementation.

The platform will offer researchers and clinical trials a DNA methylation array analysis service including bioinformatic pipelines for data normalization and filtering, quality control, and the use of existing CpG site algorithms for predicting e.g. subgroups or prognosis, blood cell composition, and biological age of the samples, as well as exploratory analysis for identification of new biomarkers. For example, a panel for CpG Island Methylator Phenotype (CIMP) classification has been identified as one promising prognostic marker in childhood T-cell acute lymphoblastic leukemia, and the CG Umeå node will provide a service for CIMP classification of Swedish and European samples.

By coordinating research and clinical trial DNA methylation array analysis of hematological malignancies at CG in Umeå, the node will continuously build specialized knowledge of these rare diseases and offer up-to-date bioinformatic technologies. The CG node will support the process of DNA methylation analysis from research to validation, and the introduction of DNA methylation biomarkers into clinical practice.

The CG node aims to specialize in clinical and research studies focusing on hematological malignancies by offering suited bioinformatic support, which will complement the currently established platforms for methylation array analysis in Sweden (e.g the SNP/Seq platform in Uppsala). The establishment will also expand the overall availability of an array scanner for researchers in Northern Sweden.

Users: The CG node in Umeå will offer DNA methylation array analysis on the new iScan Dx instrument including cutting-edge bioinformatic support customized for hematological malignancies. The instrument will allow CG Umeå to cater to research needs in the whole of Sweden but also European customers, e.g. the European ALLTogehter consortium, and allow for a complete analysis service. It will also allow for continued development of the platform to be able to provide close support to local researchers at Umeå University. Similar to CG Stockholm, which has grown substantially by providing genomic sequencing services for healthcare, the instrument will also allow for an increase in user fees by also performing contracted analyses for Region Västerbotten and other healthcare providers.

Finally, researchers in Sweden can use the platform to explore the large blood biobanks in Northern Sweden, both for epigenetics and genotyping analysis. The NHDS (Northern Sweden health and disease study) with > 135 000 individuals (https://www.umu.se/enheten-for-biobanksforskning/provsamlingar-och-register/northern-sweden-health-and-disease-study/), UCAN (cancer blood and tissues), and Biobanken Norr have thousands of samples that could be analyzed to explore disease biology. The new methylation screening arrays especially made for discovery studies can analyse 48 samples in one chip and will be excellent for biobank studies.

Co-founding: We have applied for 50% co-founding for the instruments within the ALF system and Barncancerfonden. Moreover, we have applied for additional funding from local infrastructure grants at Umeå University for full financing of the instrument, including overhead costs, service costs, and personnel. On these applications we await decisions.